

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 3. Pathologic Allelic Variants in *SEPN1* Associated with Multiminicore Disease

Nucleotide Change	Amino Acid Change	Exon
c.-19/+73del	Unknown	1
c.1A>G	Unknown	1
c.1AinsT	Unknown	1
c.22dup10bp	Frameshift at Q8	1
c.80dup20bp	Frameshift at R27	1
c.713-714insA	Frameshift at N238	5
c.817G>A	G273E	6
c.878A>G	H293R	7
c.943G>A	G315S	7
c.1019A>T	N340I	8
c.1315C>T	R439X	10
c.1358G>C	W453S	10
c.1384T>G	U462G	10
c.1385G>A	U462X	10
c.1397G>A	R466Q	11
c.1446delC	Frameshift at L482	11
g.17195T>C	SECIS element	3'UTR